Newborn Screening Quality Assurance Program

PROFICIENCY TESTING

Sickle Cell Disease and Other Hemoglobinopathies

Volume 15, No. 1 | Quarter 1 | February 2005

INTRODUCTION

On January 10, 2005, we distributed to all active participants the Quarter 1 proficiency testing (PT) panel consisting of five dried-blood-spot (DBS) specimens for sickle cell disease and other hemoglobinopathies. A total of 72 PT panels were mailed by overnight FedEx mail. The packages went to 54 domestic laboratories and 18 foreign laboratories. The specimen panel consisted of five DBS specimens prepared from umbilical cord blood. This PT report is a compilation of all data reports for hemoglobinopathy testing received from participants by the designated deadline date. We distribute this quarterly report to all participants, state laboratory directors, and to program colleagues by request. We received data reports from 59 newborn screening laboratories. There were 13 laboratories that did not report this quarter. We requested that participants assay all survey specimens by the analytic schemes they routinely use and report for each specimen the presumptive phenotype, the presumptive clinical assessment, and any other clinical classifications that they deem consistent with their analytic results and program operations.❖

PARTICIPANTS' RESULTS

The certification report listing hemoglobins (Hbs) by phenotype and their presumptive clinical assessments appears on page 2.

The frequency distribution of reported phenotypes and presumptive clinical assessments appears on page 3.

The individual data verification for each laboratory with evaluation comments appears on page 4.❖

The NSQAP will ship next quarter's PT specimens on April 4, 2005.❖

SPOTLIGHT

DURHAM, N.C., Jan 31, 2005 (United Press International via COMTEX) -- U.S. researchers said they have developed a new understanding of the causes for symptoms of sickle cell disease. Researchers at Duke University Medical Center and elsewhere said they have found that an inability of red blood cells to relax blood vessels through the release of nitric oxide is a major factor behind the disease which affects about one in every 600 African-Americans. They also found that differences among patients' ability to process nitric acid in the blood correspond to the severity of their disease. Because of the discovery, new therapies to restore nitric oxide to blood cells might be an effective way to treat the disease. The researchers plan to re-evaluate the role nitric oxide may play in other blood diseases and conditions characterized by a deficiency in oxygen delivery to tissues, such as heart failure, diabetes and hypertension.

The findings appear in the Jan. 31, 2005 edition of Proceedings of the National Academy of Sciences.❖

ACKNOWLEDGMENTS

The specimens for this survey were prepared from umbilical cord blood samples supplied by state public health laboratory in Alabama.❖

CDC/APHL

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CDC
CENTERS FOR DISEASE CONTROL AND PREVENTION

Newborn Screening Quality Assurance Program
Sickle Cell Disease and Other Hemoglobinopathies
Frequency Distributions

Year: 2005 Quarter: 1

Phenotypes

Specimen Number	Hemoglobin Phenotypes	Frequency Distributions
1531	FAC	52
	FCA	7
1532	FA	59
1533	FAS	59
1534	FA	59
1535	FCS	2
	FSC	56
	SC	1

Clinical Assessments

Specimen Number	Presumptive Assessments	Frequency Distributions
1531	03 HbC carrier	58
	15 β Thal with other	1
1532	01 Normal	59
1533	02 HbS carrier	59
1534	01 Normal	59
1535	05 Sickle-Hb C disease	59

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Sickle Cell Disease and Other Hemoglobinopathies
Specimen Certification Report

Year: 2005 Quarter: 1

Presumptive Clinical Phenotypes

	Specimen 1531	Specimen 1532	Specimen 1533	Specimen 1534	Specimen 1535
Expected Presumptive Phenotype	FAC	FA	FAS	FA	FSC
Accepted Presumptive Phenotypes					

Presumptive Clinical Assessments

	Specimen 1531	Specimen 1532	Specimen 1533	Specimen 1534	Specimen 1535
Expected Presumptive Clinical Assessment	03	01	02	01	05
Accepted Presumptive Clinical Assessments	21,15	21	21,13, or 15	21	21

01 Normal--no abnormal Hbs found
02 Hb S carrier
03 Hb C carrier
04 Sickle cell anemia
05 Sickle-Hb C disease
06 Sickle-Hb D disease
07 Sickle-Hb O disease
08 Hb D carrier
09 Hb E carrier
10 Hb G carrier
11 Hb O carrier
12 Hereditary persistence of HbF
13 Sickle β thalassemia
14 Hb E β-thalassemia
15 β-thalassemia with HB C, S, or other variant
16 α-thalassemia (Hb Barts)
17 Transfused infant
18 Hb E homozygote
19 Combination of more than one of the above
20 Assessment is not listed, please specify
21 Unsatisfactory specimen, please describe
22 Unidentified variant carrier
23 Hb E-alpha thalassemia

(NE) Specimen not evaluated. For educational purposes only.